

**Klinefelter's syndrome linking awareness with genetic insights**

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Madam, Klinefelter Syndrome is a genetic condition affecting males resulting in an additional X chromosome leading to multiple variations, including 47, XXY, 48, XXXY and 49, XXXXY.<sup>1</sup> It is linked to chromosome non-disjunction that can happen during anaphase's meiosis I, II or mitosis, leading to the presence of an extra X chromosome.<sup>1</sup> Depending on genetic penetrance, Klinefelter's syndrome leads to a wide range of phenotypes, the hallmarks being gynaecomastia, hypergonadotropic hypogonadotropism and azoospermia.<sup>2</sup>

There are 250,000 men born with Klinefelter syndrome in the United States out of a male population of 148 million, and the condition affects 3% to 4% of male births.<sup>3</sup> Nevertheless, despite increased awareness and concern for people born with it, research on this hereditary condition in Pakistan remains limited. Increasing public awareness of KS beyond medical circles is crucial to ensure individuals receive the resources they need.

This condition presents as a male with effeminate features such as gynaecomastia, and underdeveloped genitalia.<sup>2</sup> Additionally, the patient may suffer from infertility, cardiovascular symptoms, cerebrovascular symptoms, metabolic syndrome and an increased risk of other comorbidities.<sup>4</sup> Cardiovascular mortality is higher in those with Klinefelter's Syndrome due to aortic valve anomalies, pulmonary embolism, peripheral vascular disease, and deep vein thrombosis.<sup>4</sup>

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Fortunately, a lot has changed in terms of therapy possibilities. Some males with KS can father children by testicular sperm extraction (TESE).<sup>5</sup> According to studies, collecting sperm using TESE has a 40% success rate, and employing an intracytoplasmic sperm injection (ICSI) technique results in a 43% pregnancy rate per cycle.<sup>5</sup> Many of the symptoms and effects of Klinefelter syndrome can be managed, and in some cases, even prevented with proper treatment, such as hormone therapy period.<sup>4</sup> With the right care, people with Klinefelter's syndrome can safely become parents and lessen the impact the condition has on their day-to-day activities.<sup>4,5</sup>

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**KK, MF & SSN:** Concept, design, data acquisition, analysis,

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